Population-Specific and Founder Mutations in Dysferlin

Population studied: Libyan Jewish
Patients analyzed: 29 patients from 12 families
Specific mutation(s): Original numbering: **1624delG** (single base deletion at codon 1624)
New numbering: Nucleotide change: **4872delG fsX9**
Protein change: **Glu1624Asp**
Carrier frequency: 10% of Libyan Jews

Reference:
Argov Z, Sadeh M, Mazor K, Soffer D, Kahana E, Eisenberg I, Mitrani-Rosenbaum S, Richard I, Beckmann J, Keers S, Bashir R, Bushby K, Rosenmann H. 2000. Muscular dystrophy due to dysferlin deficiency in Libyan Jews. Clinical and genetic features. *Brain* 123(6):1229-37.

Population studied: Spanish (town of Sueca, Spain)
Patients analyzed: 8 patients from 5 families
Specific mutation(s): Nucleotide change: **C6086T**
Protein change: **R1905X**
Carrier frequency: 2% of Sueca residents

Reference:
Vilchez JJ, Gallano P, Gallardo E, Lasa A, Rojas-Garcia R, Freixas A, De Luna N, Calafell F, Sevilla T, Mayordomo F, Baiget M, Illa I. 2005. Identification of a novel founder mutation in the DYSF gene causing clinical variability in the Spanish population. *Arch Neurol* 62(8):1256-9.

Population studied: Italian
Patients analyzed: 3 patients from 2 families
Specific mutation(s): Nucleotide change: **C2875T**
Protein change: **R959W**
Carrier frequency: unknown

Reference:
Cagliani R, Fortunato F, Giorda R, Rodolico C, Bonaglia MC, Sironi M, D'Angelo MG, Prelle A, Locatelli F, Toscano A, Bresolin N, Comi GP. 2003. Molecular analysis of LGMD-2B and MM patients: identification of novel DYSF mutations and possible founder effect in the Italian population. *Neuromuscul Disord* 13(10):788-95.

Population studied: Aboriginal Canadian
Patients analyzed: 14 patients from 3 families
Specific mutation(s): Nucleotide change: **C2745G**
Protein change: **P791R**
Carrier frequency: unknown

Reference:
Weiler T, Bashir R, Anderson LVB, Davison K, Moss JA, Britton S, Nylen E, Keers S, Vafiadaki E, Greenberg CR, Bushby KMD, Wrogemann K. 1999. Identical mutation in patients with limb girdle muscular dystrophy type 2B or Miyoshi myopathy suggests a role for modifier gene(s). *Hum Mol Genet* 8(5):871-7.

Population studied: Palestinian
Patients analyzed: 10 patients from 1 family
Specific mutation(s): 23 bp insertion (tandem duplication) at the 3' end of exon 45
Carrier frequency: unknown

Reference:
Mahjneh I, Marconi G, Bushby K, Anderson LV, Tolvanen-Mahjneh H, Somer H. 2001. Dysferlinopathy (LGMD2B): a 23-year follow-up study of 10 patients homozygous for the same frameshifting dysferlin mutations. *Neuromuscul Disord* 11(1):20-6.

Population studied: Russian
Patients analyzed: 9 patients from 1 family
Specific mutation(s): Nucleotide change: **TG573/574AT**
Protein change: **V69D**
Carrier frequency: unknown

Reference:
Illarioshkin SN, Ivanova-Smolenskaya IA, Greenberg CR, Nylen E, Sukhorukov VS, Poleshchuk VV, Markova ED, Wrogemann K. 2000. Identical dysferlin mutation in limb-girdle muscular dystrophy type 2B and distal myopathy. Neurology 55(12):1931-3.